

FIGURE 1

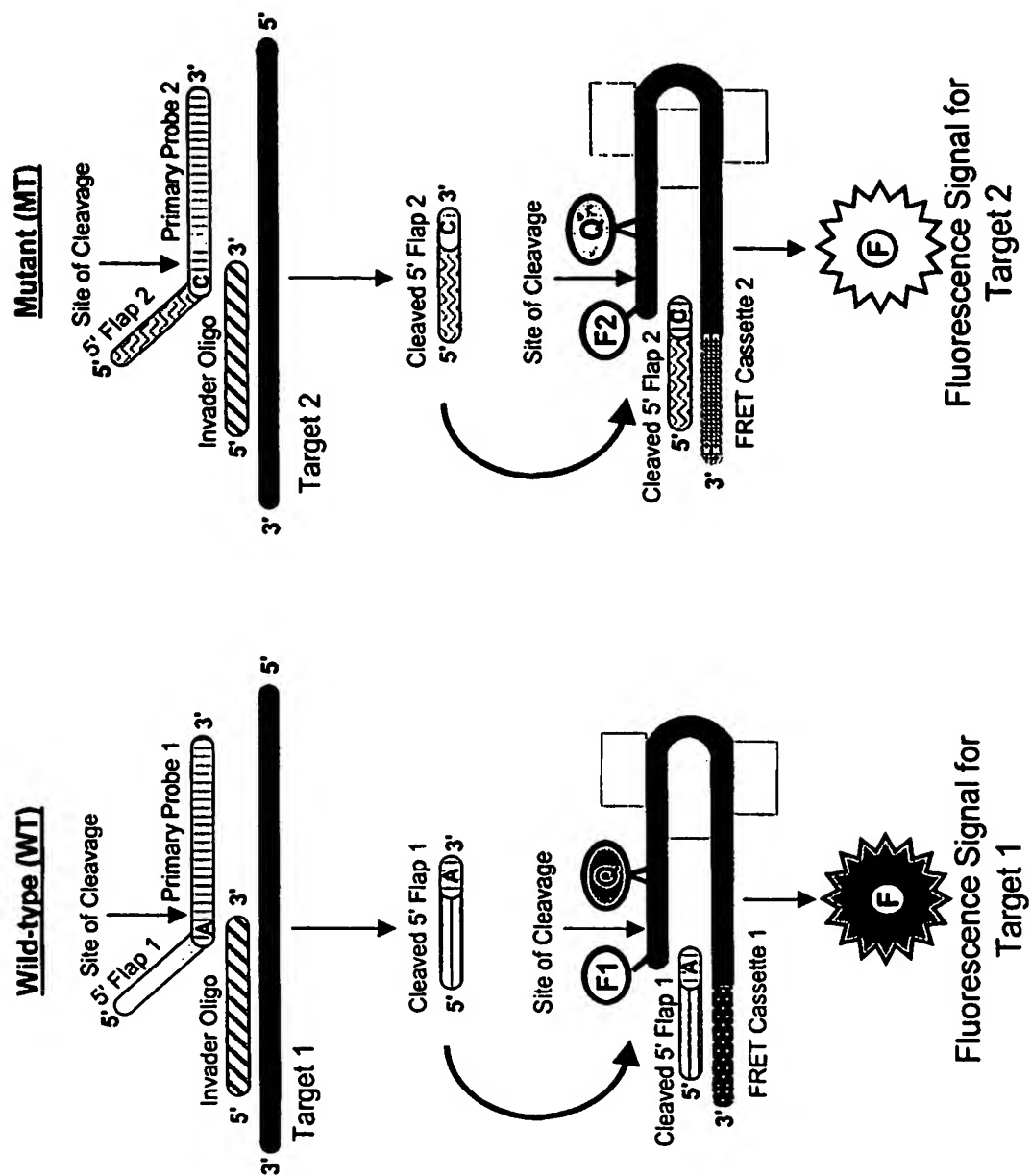


FIGURE 2

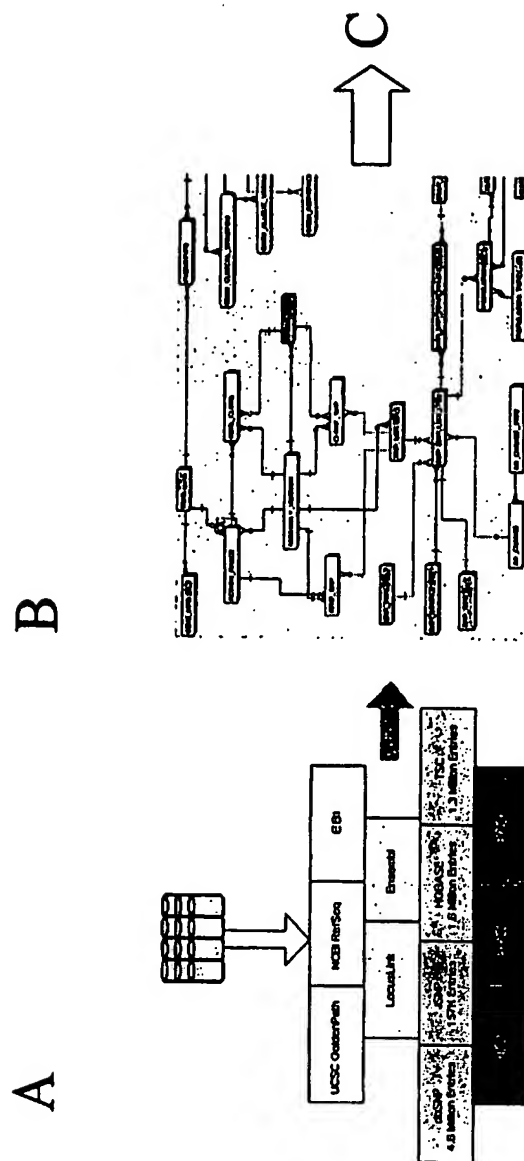
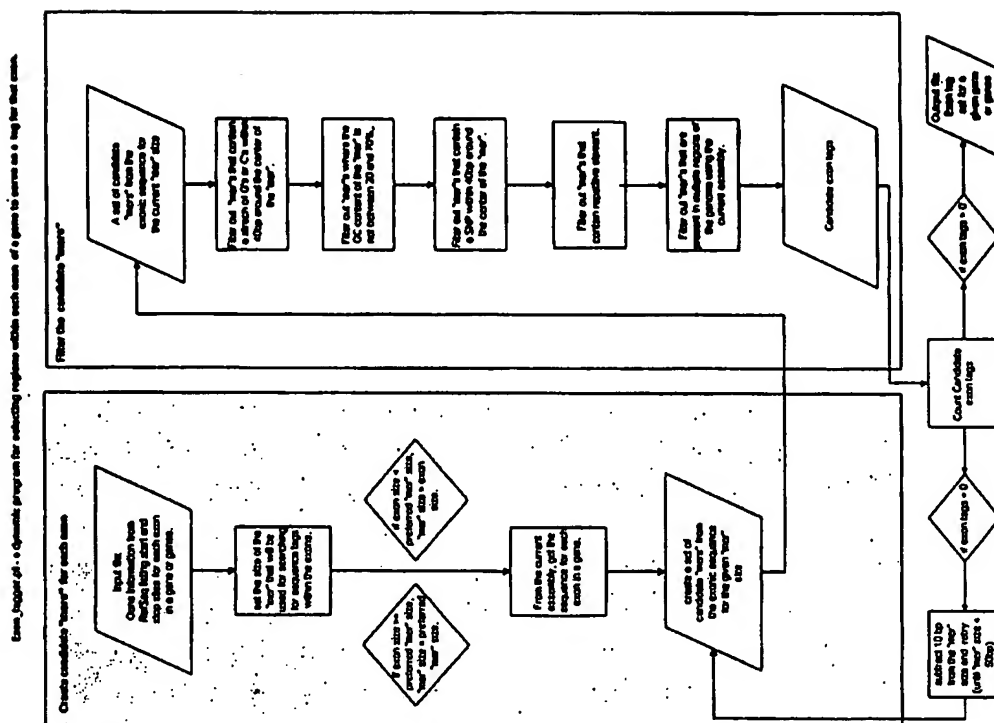


FIGURE 2 (continued)



U

↑  
B

Figure 3

Chrom	Gene	%GC	Probe Oligo Sequence	SEQ ID NO	Invader Oligo Sequence	SEQ ID NO
1	ACTA1	N/A	ACGGACCGGGAGGAGCCCTGTGACAT	1	1CCATCCAGGGAAGAGTGGCCTGTTT	100
1	ACTA1	N/A	ACGGACCGGGAGGAGCCCTGTGACATT	2	2CCATCCAGGGAAGAGTGGCCTGTTT	101
1	ACTA1	N/A	ACGGACCGGGAGGAGCCCTGTGACATT	3	3CCATCCAGGGAAGAGTGGCCTGTTT	102
1	ACTA1	N/A	ACGGACCGGGAGGAGCCCTGTGACATTTC	4	4CCATCCAGGGAAGAGTGGCCTGTTT	103
1	ACTA1	N/A	ACGGACCGGGAGGAGCCCTGTGAGAAC	5	5CGTTGGCACCCCATCCAGGGAAGAT	104
1	ACTA1	54%	ACGGACCGGGAGGAGCCCGCAGTCACT	6	6GGCGGCTGAGCTCCAGCCAT	105
1	ACTA1	58%	ACGGACCGGGAGGAGGTGGAGTGTG	7	7TTCGTGCTCTGAGAAAGTCGCGTGCC	106
1	ACTA1	63%	ACGGACCGGGAGGAGGATCTCACCA	8	8GGCCTGGACCTGGCGGGCT	107
1	ACTA1	62%	ACGGACCGGGAGGAGGATCTGCGACA	9	9GCCGCTTTCACGAGGCCGGAT	108
1	HIST2H2BE	58%	ACGGACCGGGAGGAGGATGACCGGACACA	10	10GGCCCATCGCACGGCAGCAACTT	109
1	HIST2H2BE	56%	ACGGACCGGGAGGAGTGGTGACAGCTC	11	11ACAATTACGCCCGGGTTCCGCAACAA	110
1	SV2A	59%	ACGGACCGGGAGGAGGAGGAGCTGAG	12	12CAGGGCCAGAGAGCTGCCAAGGG	111
1	CRA	59%	ACGGACCGGGAGGAGTCCCTGCTGGCA	13	13TCAGCTCTGCTTGGCGGGACAGTCC	112
21	DSCR8	60%	CGCGCCGAGGCAAGAAATTCATGTCTCAG	14	14CACTGCAGCTCCACCTCCCAGTTT	113
21	DSCR6	52	CGCGCCGAGGCTCGACTCACGGCA	15	15CCAGGCTGGAATGCAATGGTGCGATT	114
21	DSCR8	58	CGCGCCGAGGCTCGAGTGTCTGAGAT	16	16ACTGGAGGTGGAGGCTGCAGTGATT	115
21	DSCR1	56	CGCGCCGAGGCTCGAGTGTGAGAGA	17	17GGCCTCCAGTCTGGTGCAGCTCAGCATT	116
21	AML1	58%	CGCGCCGAGGCTCGACCTGTGCGA	18	18GGTCTGTCTGTGCCAAGGGCAGGTTGATT	117
21	AML1	56%	CGCGCCGAGGCTCTCTGCAGAACT	20	20GCCTCAGTGGAGACAAGTGGGAAACATGTT	119
21	AML1	54%	CGCGCCGAGGCTACACAGAGCA	21	21TGGACGTGCCAGCGGCATGACAT	120
21	DSCR9	57%	CGCGCCGAGGAGTCTGTAACTG	22	22CTTCAAAACCCACCGCAAGTCGCCAT	121
21	DSCR9	57%	CGCGCCGAGGAGGCGGAGCACTG	23	23CTCCACCCGTTCATCCACGGCT	122
X	L1CAM	58%	CGCGCCGAGGCTGAGCAACGTGC	24	24GCACCTCCACCCGTTCATCCCT	123
X	L1CAM	58%	CGCGCCGAGGCTGAGCAACGTGCA	25	25CGGATTCAGCGTGGCGCCCTGATT	124
X	L1CAM	58%	CGCGCCGAGGCTGAGTGCAGCAG	26	26CGGATTCAGCGTGGCGCCCTGATT	125
X	L1CAM	58%	CGCGCCGAGGCTGAGTGCAGCAG	27	27CGGATTCAGCGTGGCGCCCTGATCT	126
X	L1CAM	58%	CGCGCCGAGGCTGAGCAACGTG	28	28CCGATTCAGCGTGGCGCCCTT	127
X	L1CAM	58%	CGCGCCGAGGAGCAACGTGCAGC	29	29GATTCAGCGTGGCGCCCTGATCCTT	128
X	L1CAM	58%	CGCGCCGAGGAGTGCAGCCAGTGA	30	30CCGATTCAGCGTGGCGCCCTT	129
X	PDCD8	52%	CGCGCCGAGGCTACAGCAGCTC	31	31CAGATTTTGGTGGCTTCGGGTAATGCAGAT	130
X	PPEF1	53%	CGCGCCGAGGCTGACGCGAAGTG	32	32TGCTGGCCTTCAGTCGAGCTTTGTAACCTT	131
Y	SRY	55%	CGCGCCGAGGCTGACGCGAAGTG	33	33GCTTCCCGCAGATCCCGCTTCGGTAT	132
Y	EIF1AY	56%	CGCGCCGAGGCTTAAGCGGAGCTGA	34	34GACCTCTCCGACTCCTTCTGGCGGTTACTAT	133
18	GATA6	54%	CGCGCCGAGGCTGCTTCTGCG	35	35CACAGCATTCACACGGGTTACCCCTT	134
18	SERPINB2	57%	CGCGCCGAGGCTGCTTCTGCTGCA	36	36CTGCCACAACTGTGGGCTCCATGTT	135
13	DLEU1	57%	CGCGCCGAGGCTTCTGCGATGTGC	37	37AGGAGAGCGGTGCACACGATGAC	136
13	ABCC4	56%	CGCGCCGAGGAGGAGCAGCTAGGTG	38	38CGGCTGGCTGTGATCAGACTGCCGT	137
13	POU4F1	65%	CGCGCCGAGGCTTCTGAGGTTG	39	39CGTGGGCTCAGTCCAGCAGAGCAT	138
13	POU4F1	65%	CGCGCCGAGGCTCAGCTCCGAGG	40	40CTGACACAAAGTACCCGTCGCTGCAT	139
13	POU4F1	63%	CGCGCCGAGGCTCAGCTCCGAGG	41	41CCTGCTCCGAGAAAGATGCCGCCCATCT	140
13	POU4F1	54%	CGCGCCGAGGCTCAGCTCCGAGG	42	42CCACTCACTTCCCGGATTCGAGAGCAT	141

## Aneuploidy Probe and Invader Designs

13	PCDH9	CGCGCCGAGGCACTGTCACTGCAGC	57%	43	ACTACCCACCGGCTCATGGTCTCCTAGACT	142
13	PCDH9	CGCGCCGAGGCTGCACCTCTGAGGCA	54%	44	GGCCTTTGCTTGAAGCCTCTTGGGAAT	143
13	PCDH9	CGCGCCGAGGCACTCTCTGCTCGA	54%	45	ACAGTGTGGTGTAGTGACGGCAGCTT	144
13	PCDH9	CGCGCCGAGGCTCGTGGTAGAGAA	51%	46	GTGCCCTCTCAGCCATTCTTGCC	145
18	FLJ23403	CGCGCCGAGGCTGGATCTGTGTGG	52%	47	TGGACGGACAACTTTGAGCCTGTCCAT	146
18	KIA0222	CGCGCCGAGGCTGGAGACCAGATCA	56%	48	CGAAGAGGTGACTTCGACCGAGCTCTCCAT	147
21	CLDN17	CGCGCCGAGGCATGCACAGATGCC	56%	49	CGTAGAGCTGTGCACCTGACCTGCTTT	148
21	MGC33285	CGCGCCGAGGCATGCTTCGGAGACTC	56%	50	CTCCAGTCAGGCTCCACCAGCT	149
21	PCP4	CGCGCCGAGGCTGGTGCATCCATGTC	52%	51	GGCCACCGCTGCACGTTCTGTCTT	150
X	PFKFB1	CGCGCCGAGGCTGCTCCCTGAAGGT	56%	52	TGGCCAATTCATTAGTCCACGGGCATT	151
X	PFKFB1	CGCGCCGAGGCTGCTCCCTGAAGGT	56%	53	TGGCCAATTCATTAGTCCACGGGCATT	152
X	PFKFB1	CGCGCCGAGGCTGCTCCCTGAAGGT	56%	54	GTGACTGGTCCACACCTTCAGGGAGCTT	153
X	ZNF157	CGCGCCGAGGCTGCTCCCTGAAGGT	60%	55	GTTACCCCTGGGCTCAGCCCAT	154
X	FLJ22843	CGCGCCGAGGCTGCTCCCTGAAGGT	62%	56	CTGCTCTGCTCCGGGCAACT	155
X	DUSP21	CGCGCCGAGGCTGCTCCCTGAAGGT	59%	57	GAGCGGAACGGCTCACTCCAGCT	156
X	DUSP21	CGCGCCGAGGCTGCTCCCTGAAGGT	59%	58	GAGCGGAACGGCTCACTCCAGCT	157
X	MGC33889	CGCGCCGAGGCTGCTCCCTGAAGGT	57%	59	GGTGACTCTCTTCTGCCACAGTGGCT	158
Y	PRKY	CGCGCCGAGGCTGCTCCCTGAAGGT	48%	60	TGCTCCCACTTCCCAAGAGAACACTCTCT	159
Y	PRKY	CGCGCCGAGGCTGCTCCCTGAAGGT	55%	61	GCACCCAGAGTAGGGTGGAGTACCCCTT	160
Y	PRKY	CGCGCCGAGGCTGCTCCCTGAAGGT	53%	62	GTCCCTCGGCTCTTAATCTCTCAAACTCAGC	161
Y	TMSB4Y	CGCGCCGAGGCTGCTCCCTGAAGGT	59%	63	TCGGACTCCCTAGCACACATGCTCTT	162
Y	TMSB4Y	CGCGCCGAGGCTGCTCCCTGAAGGT	59%	64	GCACAGTCCAGTACAGAGACCCGGAT	163
21	NRIP1	CGCGCCGAGGCTGCTCCCTGAAGGT	54%	65	GCTCTGCTGTGCAGAGAGCGCACT	164
21	NRIP1	CGCGCCGAGGCTGCTCCCTGAAGGT	54%	66	GCTCTGCTGTGCAGAGAGCGCACT	165
21	HLCS	CGCGCCGAGGCTGCTCCCTGAAGGT	60%	67	TGACCCGGACGGCAACTCTCTCT	166
21	DSCR6	CGCGCCGAGGCTGCTCCCTGAAGGT	57%	68	GCCACGATTGACTTCTACGACGATGATCTAGT	167
21	DSCR9	CGCGCCGAGGCTGCTCCCTGAAGGT	58%	69	CCGTTCCATCCAGGCGAGCAGTT	168
21	DSCR3	CGCGCCGAGGCTGCTCCCTGAAGGT	55%	70	AAGTGGCTTGACGCCATCTGGATT	169
21	DSCR4	CGCGCCGAGGCTGCTCCCTGAAGGT	56%	71	GTGGAAGGTTTGTCTTGTGCCAGGCT	170
21	DSCR4	CGCGCCGAGGCTGCTCCCTGAAGGT	56%	72	GTGGAAGGTTTGTCTTGTGCCAGGCT	171
21	DSCR10	CGCGCCGAGGCTGCTCCCTGAAGGT	59%	73	TGCCGAGCTCCACCGTGTCTCAT	172
18	CN2	CGCGCCGAGGCTGCTCCCTGAAGGT	54%	74	GGAGAAGAGAGCGGAAATCAGGAGGATGGAAT	173
21	HLCS	CGCGCCGAGGCTGCTCCCTGAAGGT	64%	75	ACCAGAGCGAGGTGAAGCTCAGCGTT	174
21	HLCS	CGCGCCGAGGCTGCTCCCTGAAGGT	64%	76	TCCTGGTACCTGCAGCCACTGCTCAAT	175
21	HLCS	CGCGCCGAGGCTGCTCCCTGAAGGT	58%	77	CGCTGCTTGAAGCGCTTTCATGCT	176
21	DSCR9	CGCGCCGAGGCTGCTCCCTGAAGGT	57%	78	TCCATTTGCCAGGCGTTGTCCATCT	177
21	DSCR9	CGCGCCGAGGCTGCTCCCTGAAGGT	57%	79	TCCATTTGCCAGGCGTTGTCCATCT	178
21	DSCR3	CGCGCCGAGGCTGCTCCCTGAAGGT	57%	80	ACCACCTCATCAGCGAGAACTTCCCGCC	179
21	DSCR6	CGCGCCGAGGCTGCTCCCTGAAGGT	63%	81	GCCTCTGCACACTCAGCAGGATGCT	180
X	MTMR8	CGCGCCGAGGCTGCTCCCTGAAGGT	59%	82	CCATGCTGAGTCCCGCCACT	181
X	MGC23947	CGCGCCGAGGCTGCTCCCTGAAGGT	53%	83	CTCCTTTTCTCCCGCAGATGCTGTAACACAT	182
X	FLJ21174	CGCGCCGAGGCTGCTCCCTGAAGGT	56%	84	TGGCCTCCAGCTGGCATTGACCTT	183
X	ESX1L	CGCGCCGAGGCTGCTCCCTGAAGGT	62%	85	CCGACCGGCTTCAGCGAGTTCT	184
X	ZNF157	CGCGCCGAGGCTGCTCCCTGAAGGT	52%	86	CTGGTGATCATATGCCCCAAGGAGCTTGATCT	185

## Aneuploidy Probe and Invader Designs

X	PCTK1	57%	CGCGCCGAGGTGAAGCCTGGTCACT	87 CTGCACCTCATCCGAGGACGTGGCC	186
X	MGC33889	62%	CGCGCCGAGGTGCGTAGCTCCAG	88 GGTGACTCTCTTCTGCCACAGTGGCCC	187
21	DSCR8	57%	CGCGCCGAGGGAGCAGTGTGAACGTG	89 CTCCCCACCGTTCCATCCAGGCT	188
21	DSCR9	57%	CGCGCCGAGGAGGCGAGAGTCTG	90 GCACCTCCCCACCGTTCATCCCT	189
21	DSCR9	57%	CGCGCCGAGGCCATCCAGGCGA	91 GTCTGCGCACCTCCCACCGTTT	190
21	DSCR9	57%	CGCGCCGAGGTTCCATCCAGGCG	92 CGCTGCGCACCTCCCCACCGC	191
Y	SRV	55%	CGCGCCGAGGACTGTACGAAGCCAC	93 GAAGTGCAACTGGACAACAGGTTGTACAGGGATT	192
Y	SRV	55%	CGCGCCGAGGAGGTTGTACAGGGATGA	94 GGTACTCTGCAGCGAAGTGCAACTGGACAAC	193
Y	SRV	56%	CGCGCCGAGGGCAGATCCCGCTTC	95 TGCTGCCGAAGAAATGCAGTTTGTCTCCCT	194
Y	SRV	54%	CGCGCCGAGGGCTTCGGTACTCTGC	96 GAAGAAATGCAGTTTGTCTCCCGCAGATCCCT	195
21	CLDN17	59%	CGCGCCGAGGTGAAGCAGGTCCAGT	97 CGCCCTGCTTATTGGCATCTGTGGCAC	196
21	DSCR10	56%	CGCGCCGAGGGCATGGCTCTGCAC	98 GCTTGAAGGTGACACACAGTGACACGGGAT	197
21	MGC33295	57%	CGCGCCGAGGCCAGTCCAGGCCTCC	99 GCAAAGTAAGGAGTCGCATCCCTGCCTCT	198
21	STCH	54	CGCGCCGAGGCCACGCTTGGTGATAG	217 GCCTATGGTCTCCACAAGGCTGACGTCTTT	218
21	STCH	53%	CGCGCCGAGGGCTTGGATAGCCACTC	219 GAGAGGCCAAGAGCCTCCATCAATCCCTT	220
X	AR	56%	CGCGCCGAGGCCAGTGTCTCCGGA	221 CGACTCTGGTACGCGAGTGCCTCGTT	222
13	CCNA1	55%	CGCGCCGAGGGCGCATGCCCTTCC	223 TCCACGAGCCAGTCCACCAAGATCGTT	224
13	CCNA1	49%	CGCGCCGAGGCGTAGGAACAGCAGC	225 GTCTGTTCTGAGAGGGAAACTGCAGCTT	226
13	ING1	52%	CGCGCCGAGGTACAGGACCAATCGT	227 GCTAGGCCCGCCTTCATCCGCG	228
21	ADAMTS5	58%	CGCGCCGAGGCCAGCAGTGCAACC	229 GAATCCCGAGGACAGACCTACGATGCCACT	230

SEQ ID NO:199  
SEQ ID NO:200

FAM FRET cassette Fam-TCT-Z28-AGCCGGTTTTCCGGCTGAGACCTCGGCGCG-hex  
RED FRET cassette Red-TCT-Z28-TCGGCCCTTTTGGCCGAGAGACTCCGCGTCCGT-hex

FIGURE 4

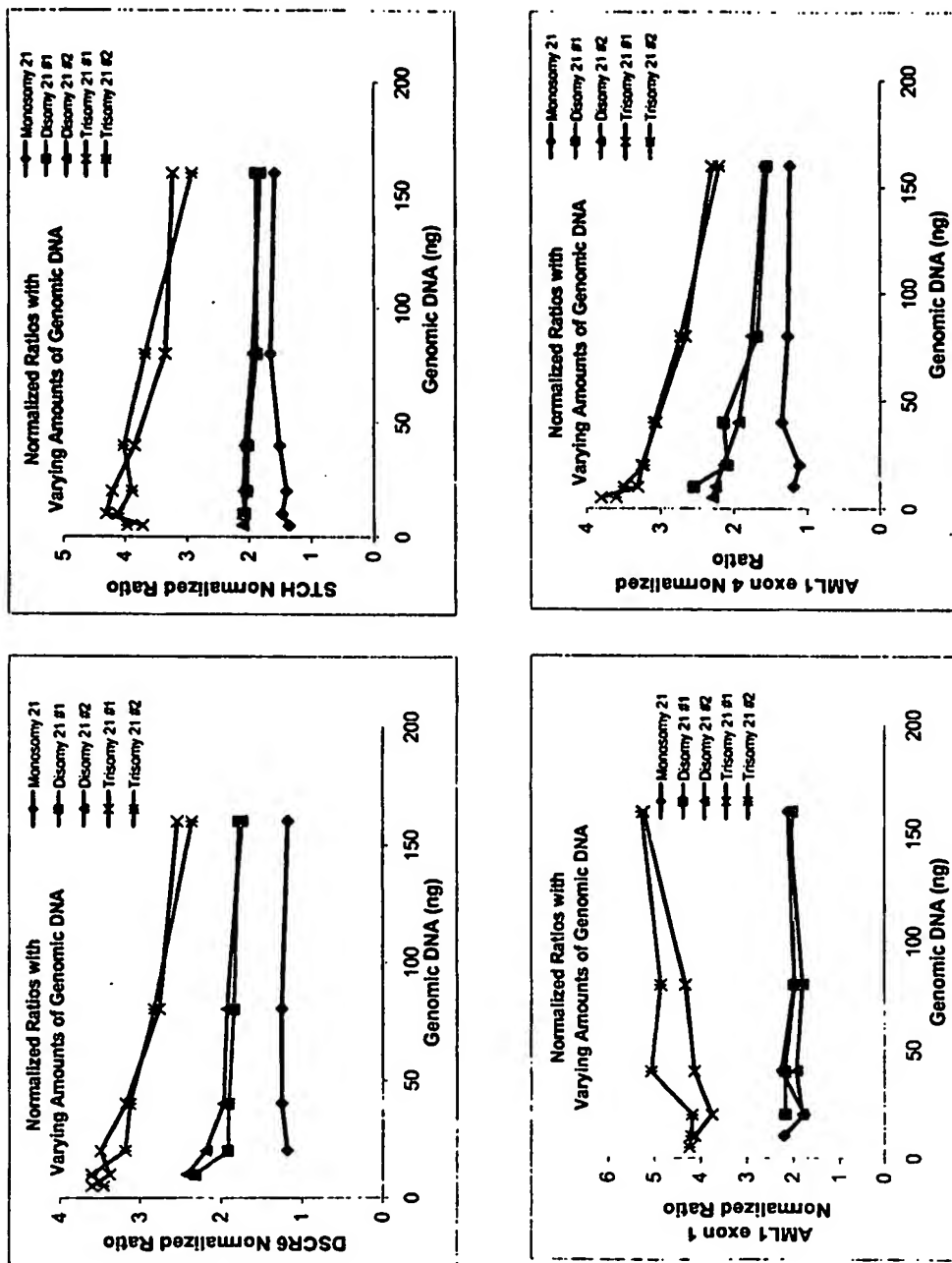
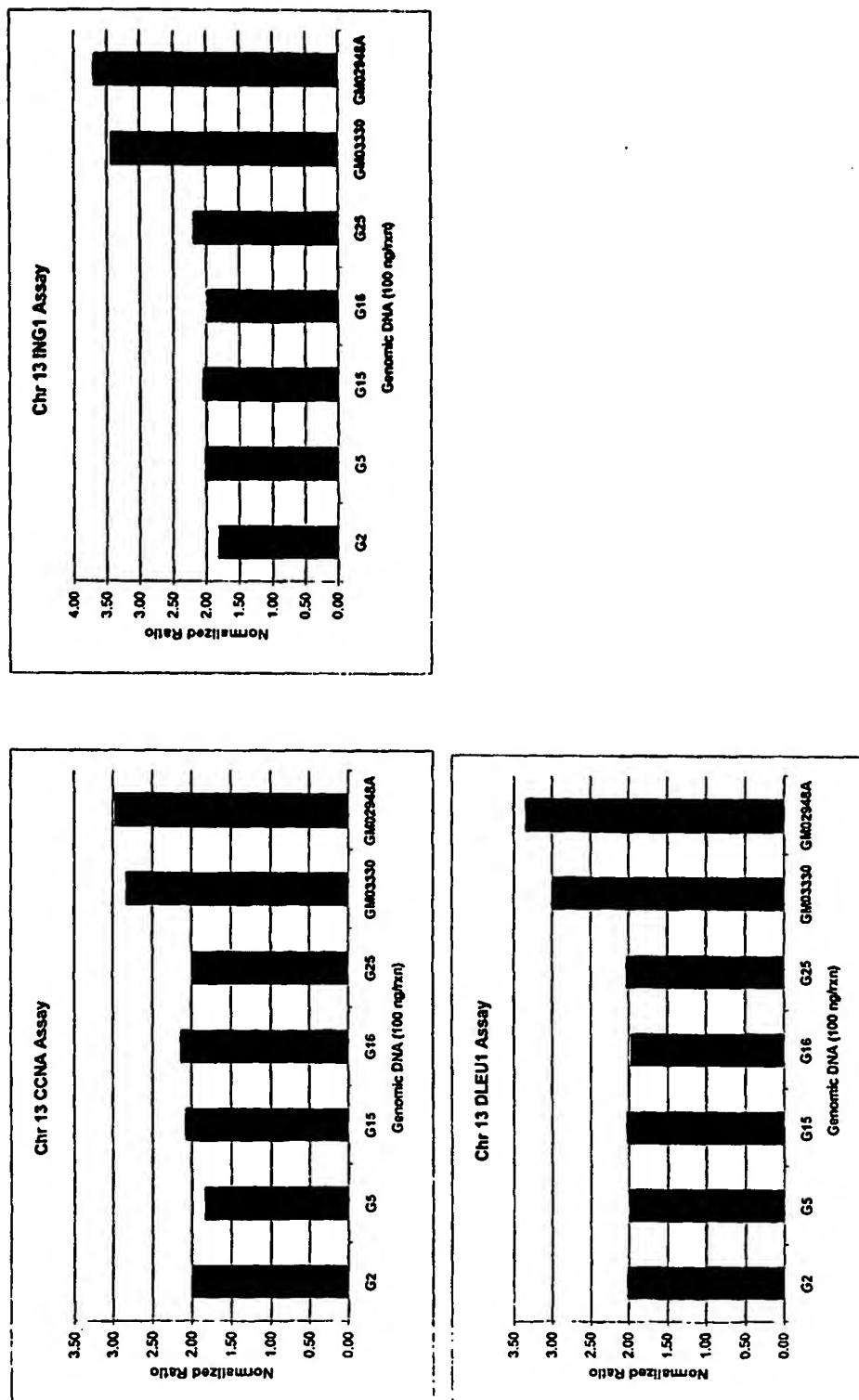


FIGURE 5





## FIGURE 6

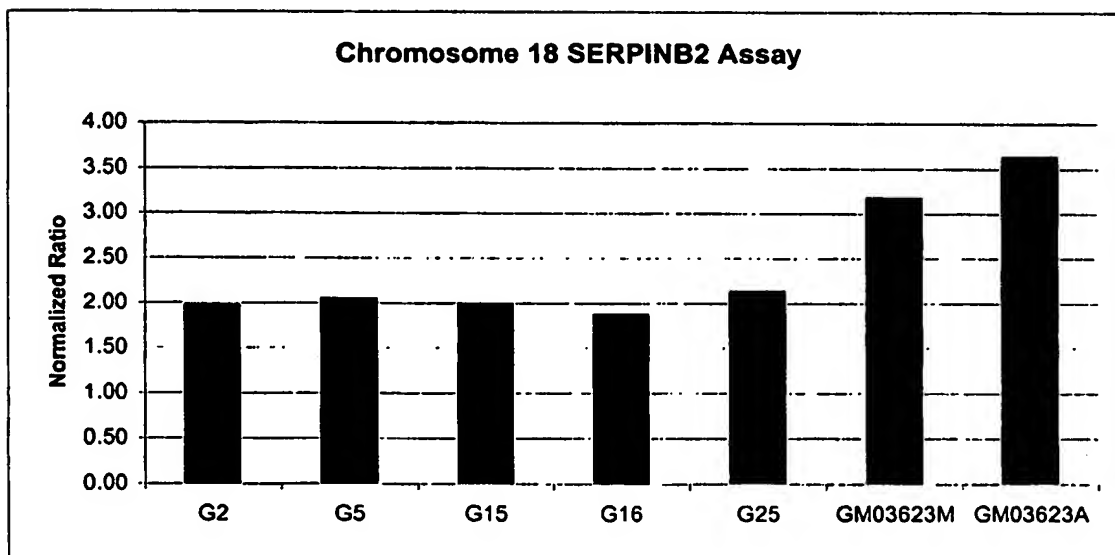
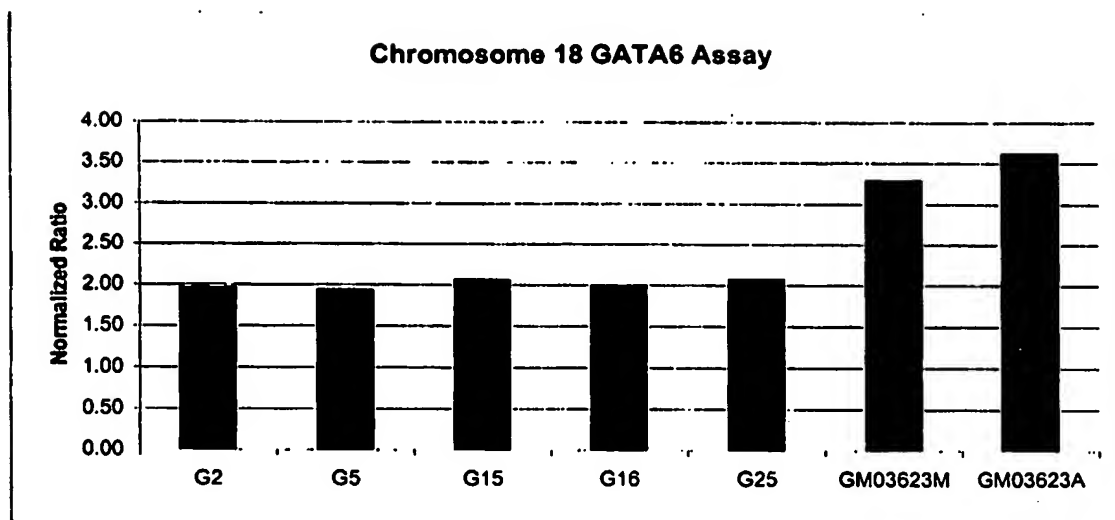


FIGURE 7

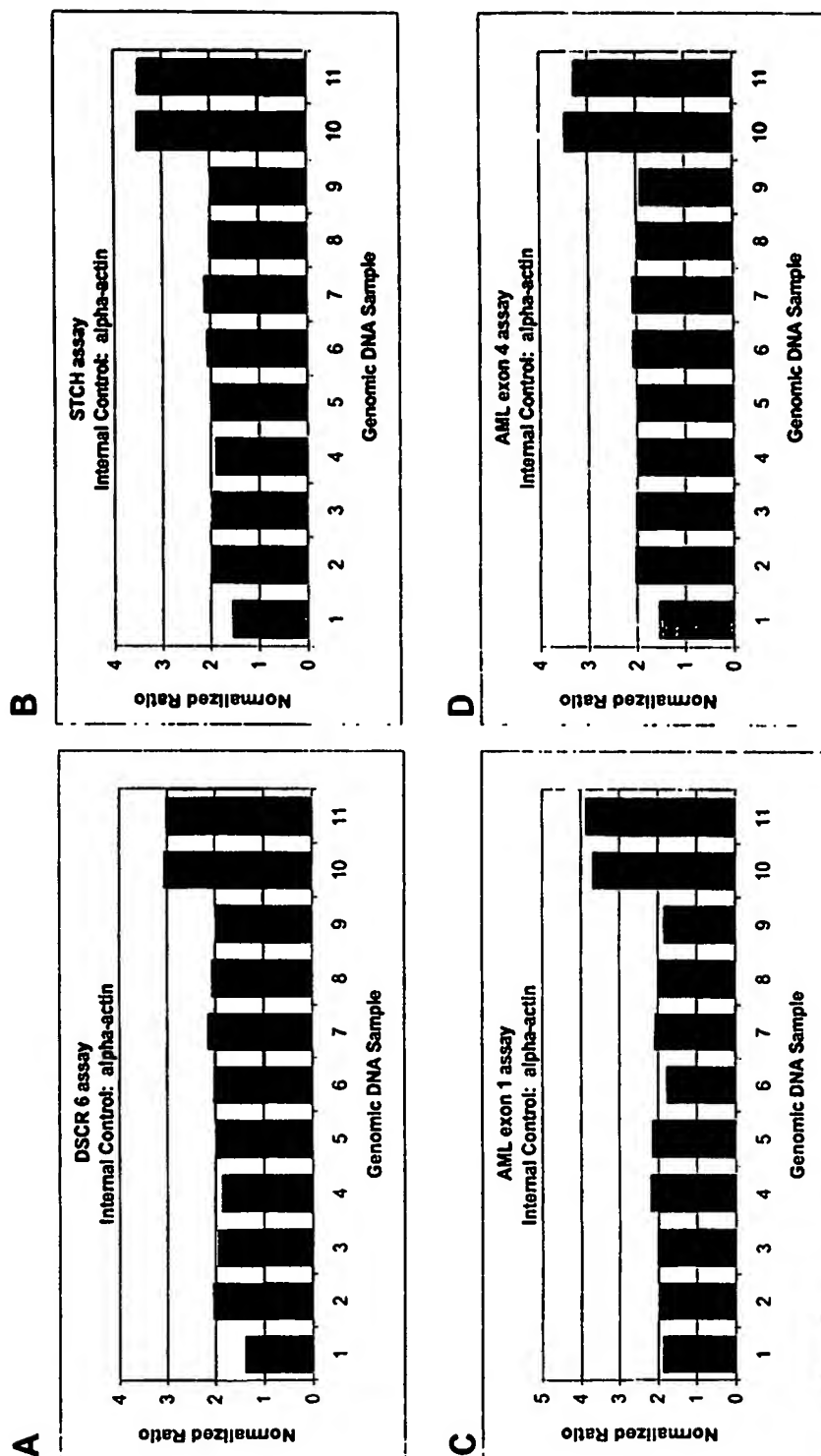


FIGURE 7 (continued)

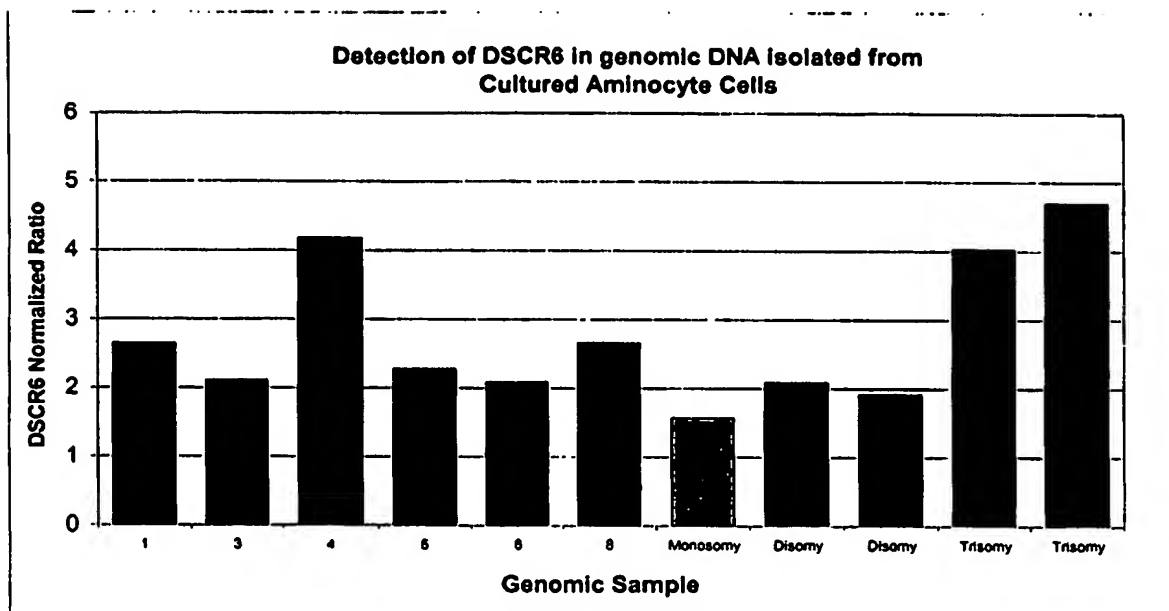
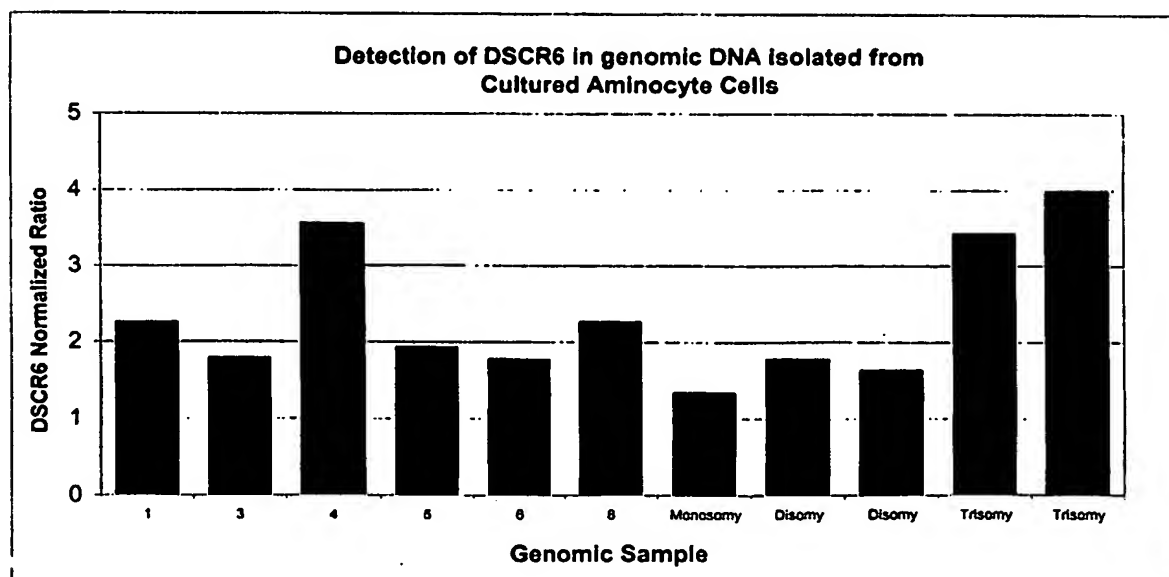
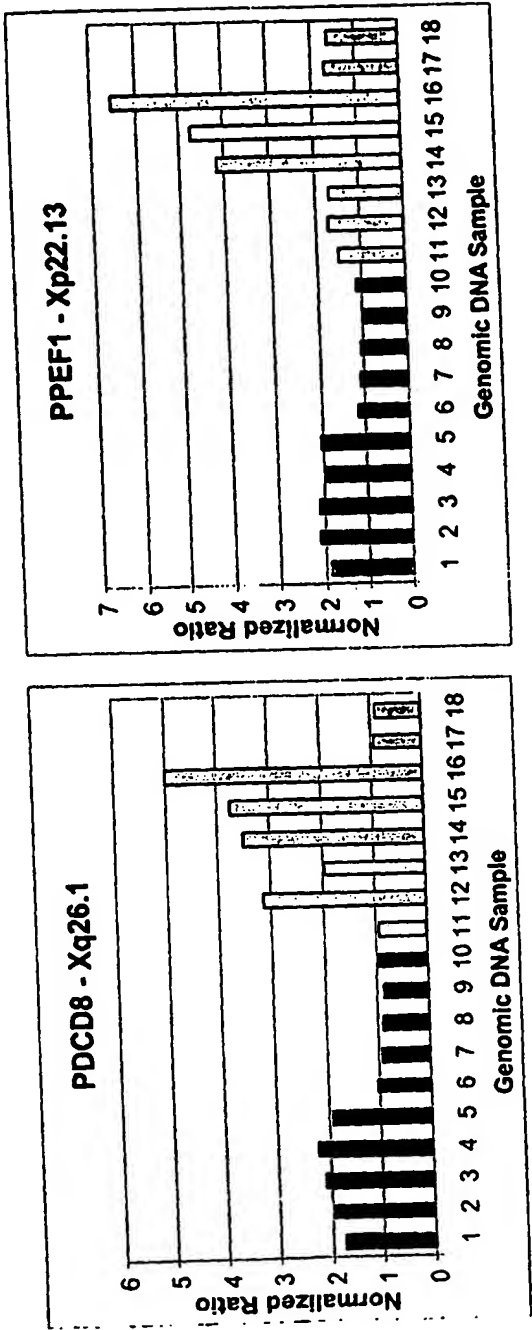
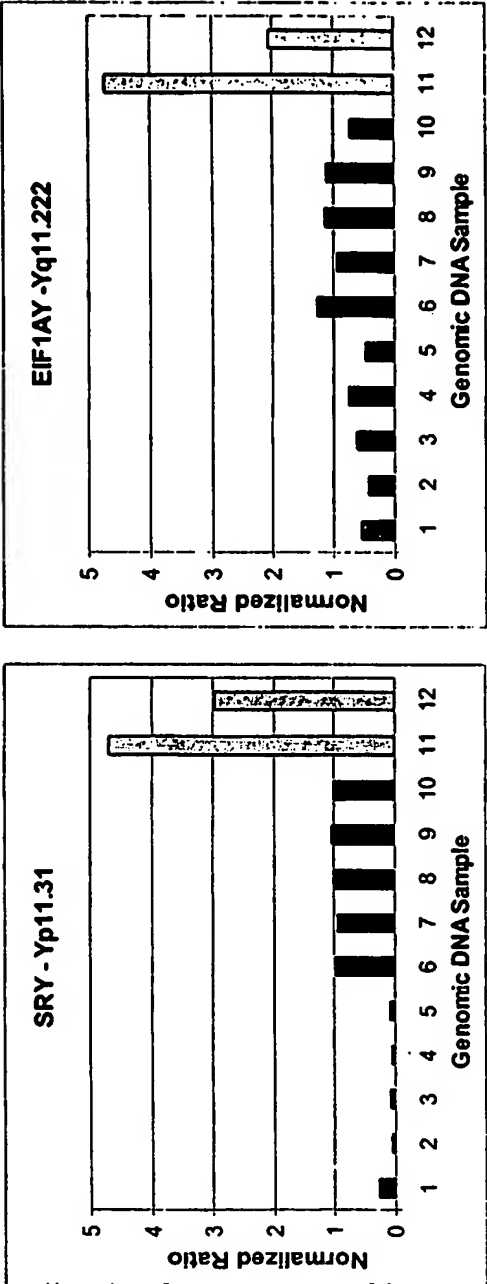
**E****F**

FIGURE 8



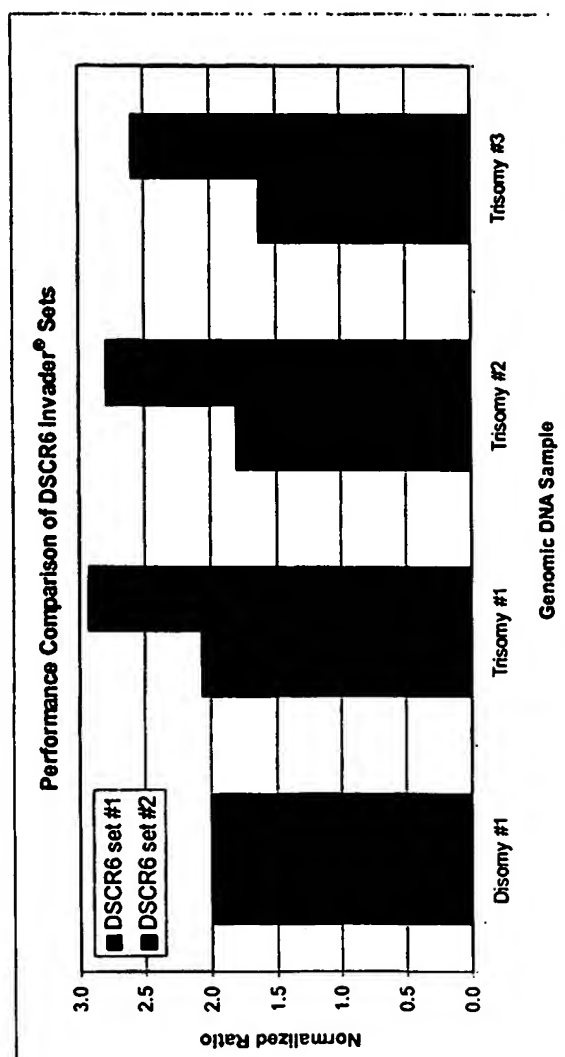
Sample #	Description
1-5	46, XX
6-10	46, XY
11	45, X
12	30% 45, X / 70% 46, X, iX (qter>cen>Qter)
13	40% 45, X / 60% 46, X, iX (qter>cen>Qter)
14	48, XXX, +18
15	48, XXXX
16	48, XXXX / 49, XXXXX
17-18	47, XYY

FIGURE 9



Sample #	Description
1-5	46, XX
6-10	46, XY
11-12	47, XYY

FIGURE 10



Trisomy #1: Coriell AG13429

Trisomy #2: Coriell AG09394

Trisomy #3: Coriell AG10098

-all genomic DNA's were prepped using the Gentra Autopure Prep

FIGURE 11A

STCH Signal (FAM)																												
% Maternal Contamination																												
ng	0	50.82	50.83	50.84	50.85	0	0	1	5	10	20	50	80	90	95	99	100	0	1	5	10	20	50	80	90	95	99	100
mg	0	0	0	0	0	50.82	100	99	95	90	80	50	20	10	5	1	0	50	49.5	47.5	45	40	25	10	5	2.5	0.5	0
Std	205	450	482	421	457	876	1024	1143	1096	1156	1122	959	788	805	749	750	765	741	785	803	809	781	639	561	553	516	497	534
	235	448	482	418	468	899	1107	1174	1157	1097	1048	940	802	765	758	725	713	803	782	821	777	722	627	578	541	478	497	509
AV	220	449	482	420	463	888	1066	1159	1127	1127	1085	950	784	765	754	738	739	772	794	812	793	752	633	570	547	497	497	522
SD	21	1	0	2	8	16	59	22	43	42	52	13	25	28	6	18	37	44	2	13	23	42	8	12	8	27	0	18
%	10%	0%	0%	1%	2%	2%	6%	2%	4%	4%	5%	1%	3%	4%	1%	2%	5%	6%	0%	2%	3%	6%	1%	2%	2%	5%	0%	3%
Signal/Bkgd		2.04	2.19	1.91	2.10	4.03	4.84	5.27	5.12	5.12	4.93	4.32	3.56	3.57	3.43	3.35	3.36	3.51	3.61	3.69	3.60	3.42	2.88	2.59	2.49	2.26	2.37	
Net Signal		229	262	200	243	668	846	939	907	907	865	730	584	565	534	518	519	552	574	592	573	532	413	350	327	277	277	302

Alpha Actin Signal (Red)																												
% Maternal Contamination																												
ng	0	50.82	50.83	50.84	50.85	0	0	1	5	10	20	50	80	90	95	99	100	0	1	5	10	20	50	80	90	95	99	100
mg	0	0	0	0	0	50.82	100	99	95	90	80	50	20	10	5	1	0	50	49.5	47.5	45	40	25	10	5	2.5	0.5	0
Std	107	309	341	299	322	381	439	436	450	490	528	553	497	562	563	586	594	284	289	323	335	342	342	334	342	345	345	368
	114	303	359	298	330	385	438	438	446	472	479	573	497	504	559	559	560	295	300	319	320	327	328	324	338	304	339	357
AV	111	306	350	298	326	383	438	437	448	481	504	563	497	533	561	573	577	290	295	321	328	335	335	329	340	325	342	373
SD	5	4	13	2	6	3	2	1	3	13	35	14	0	41	3	19	24	8	8	3	11	11	10	7	3	29	4	22
%	4%	1%	4%	1%	2%	1%	0%	0%	1%	3%	7%	3%	0%	8%	1%	3%	4%	3%	3%	1%	3%	3%	3%	2%	1%	9%	1%	6%
Signal/Bkgd		2.77	3.17	2.69	2.95	3.47	3.96	3.95	4.05	4.35	4.56	5.10	4.50	4.82	5.08	5.18	5.22	2.62	2.67	2.80	2.96	3.03	3.03	2.98	3.08	2.94	3.10	3.37
Net Signal		198	240	187	216	273	327	327	338	371	393	453	387	423	451	462	487	179	184	211	217	224	225	219	230	214	232	282
Ratio		0.59	0.55	0.54	0.57	1.23	1.30	1.44	1.35	1.23	1.11	0.81	0.73	0.87	0.59	0.56	0.56	1.55	1.57	1.41	1.33	1.19	0.92	0.80	0.72	0.65	0.60	0.56
AV	0.56																											
Normalized Ratio		2.10	1.96	1.91	2.02	4.40	4.64	5.16	4.82	4.39	3.95	2.89	2.62	2.40	2.13	2.01	2.00	5.53	5.59	5.05	4.74	4.26	3.30	2.87	2.56	2.32	2.15	2.07

FIGURE 11B

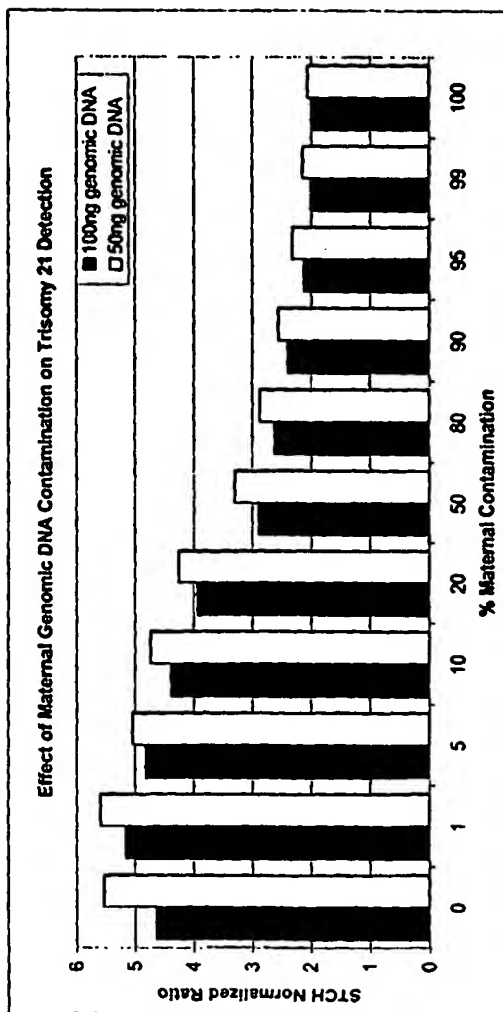
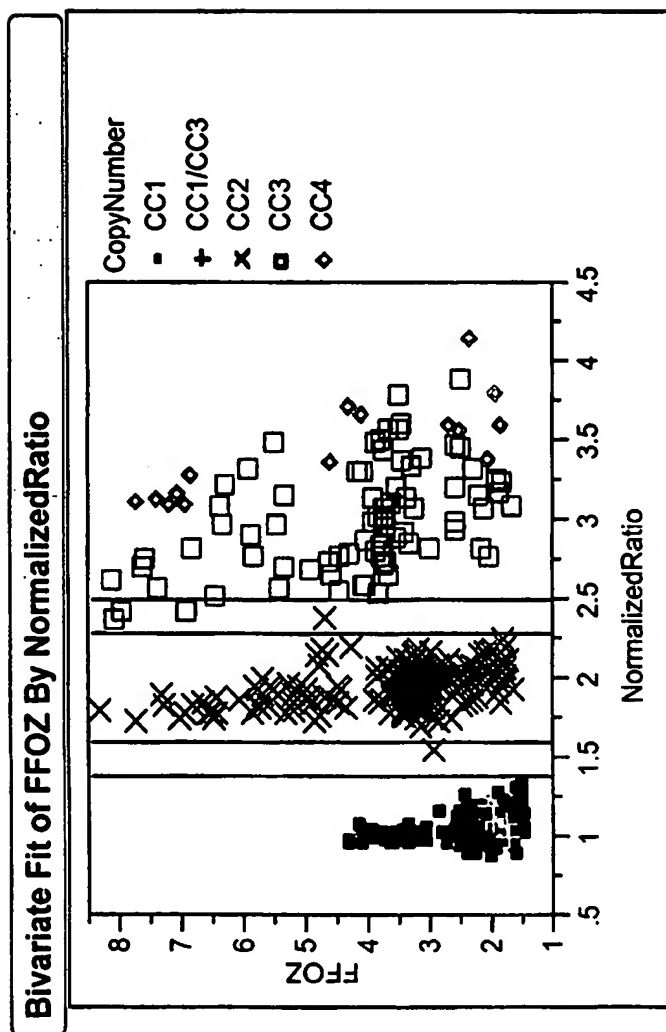




Figure 12

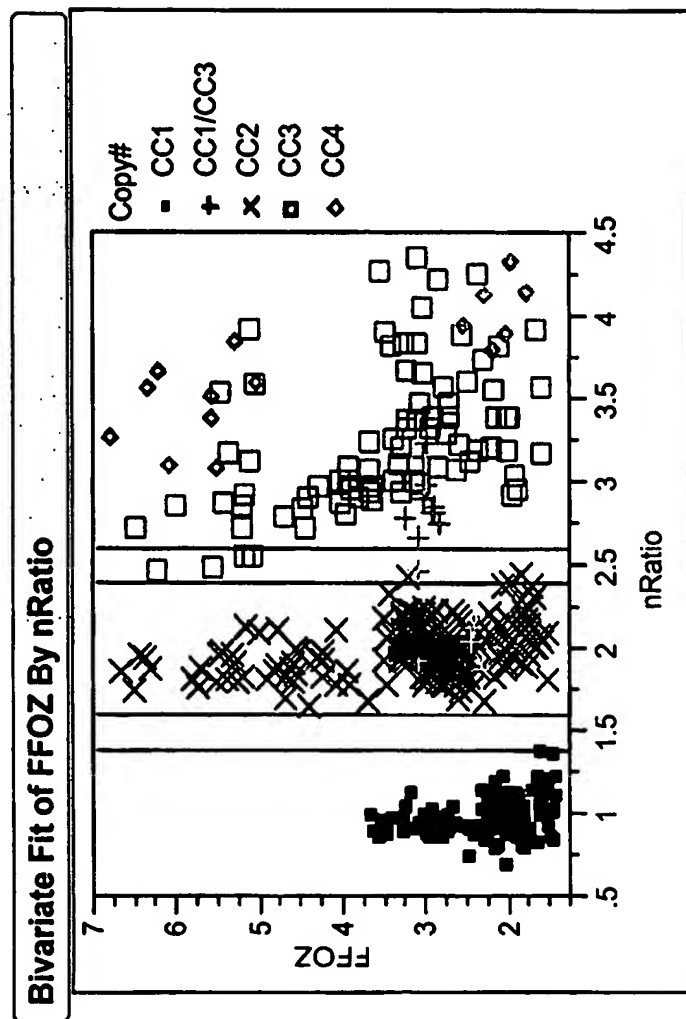
Chrom	Gene	Accession #	Cytoband	Target Sequence	SEQ ID NO:
1	alpha actin (ACTA1)	M20543	1q42.13	(ATGTCACAGGGTTCC)AACAGGCCACTCTTCCCTGGATGG)	203
1	HIST2H2B E	NM_003528	1q21.2	GTATCCACAGGA(GGCCCCCATCGCACGGCAGAACT)GJGTGACCGGACACA AAAGCTGCTGCCAAGCGTCAGTCATACAGAGCTGTACAGCAGT	204
13	DLEU1	NM_005887	13q14.2	G(GCACATGCCGCGAGAT)CATCGTGGTGCACGGCTCTCCCT)TTGCTTCTT CGGTTGCAGTCTCTTGTCTTCTTGGCGTGGTGTAGCGCTTT	205
13	PCDH9	NM_020403	13q21.33	ATGGTAACCGTTGCTCGACGAGGAGTGGAGCTGCCGTCACTAGCCAGC ACTGT)CACCTTTGTGAATGGCTGTCTCTCTCTATCTAAGGACC	206
18	FLJ23403	NM_022068	18p11.22	GCCAAACACTTCAGGATGAATATGTGAGCATAGATGTCTCT(CCACACAGATC CAGCTGGACAGGCTCAAAGTTGTCTCGTCCA)CACCCAGTC	207
X	PFKFB1	NM_002625	Xp11.21	ATGGTCTCTTCATGTGACTGGTCCACACCTTCAGGGAGCTG)ATGCC TGGGACTGAATGAAGTTGGCCA)GGGCATAGGCA	208
Y	PRKY	NM_002760	Yp11.2	CTCCTTCT(GCACCCAGAGTAGGGGTGGAGTATACCCCTT)PCCATGTGT AGTGAATATG)TGTGCAGTGAGAGGCCAGTTGGGTCCCTCGGC	209
21	NRIP1	NM_003489	21q11.2	GGTCCGATTAAAGTCTTCG(GACACTGGTAAGGCAG)GTGCGCTTCTC TGACAGCAGCAGAGC)CATACCAAGAATGGGGCACTCTTAGCAT	210
21	HLCS	NM_000411	21q22.13	GAGGATG(AGGTTTCTCAGCATGT)GAAAGGAGTTGCCGTCCGGGTGCA CAGTCACAACCTCGCCGCCCTCCTGGTGAACCTGGAGGAAGCCA	211
18	CN2	NM_018235	18q22.3	TGGCC(GGAGAAGAGAGCGGAAATCAGGAGGATGATGGAAG)TTGCTGC TGACAGAT)GTTAAGCAGTTGGGGGGCTCTGTGGAACCTGGTGGATA	212
X	MTMR8	NM_017677	Xq11.2	CCTTGGGTACCGTAATATGAT(CCATGACTGCAGTTCCCGCCAC)CGAA GATCTCAGTGT)ACTCCAGATGCCCGCCACCGGTCTAGCCG	213
X	FLJ21174	NM_024863	Xq22.2	GAAGAGTTCAAGTCTGCTAGGTTCTTATTTTGGACTCTCATGATGCT)GAG GTCAAGATGCCAGCTGGAGGCCA)GGCGCCGAGCTCACGCCCT	214
X	PCTK1	NM_033018	Xp11.3	GAGATTGTCACGAGGAGTGAAGATGGGGTCTGATGGGAGAGTGTGAC CAGGCTTCA)GCCACGCTCTCGGATGAGGTGCAG)TCTCCAGTGA	215
Y	SRY	NM_003140	Yp11.31	TCATCCCTGTACAACTGTGTCCAGTTGCACCTTCGCT(GCAGAGTACCG AAG)GGGATCTCGGGGAAGCAACTGCAATTC)TTC)SSCAGCA	216

Figure 13A  
Chromosome Xp Invader Assay:PFKFB1+PCTK1 –Varying DNA Levels



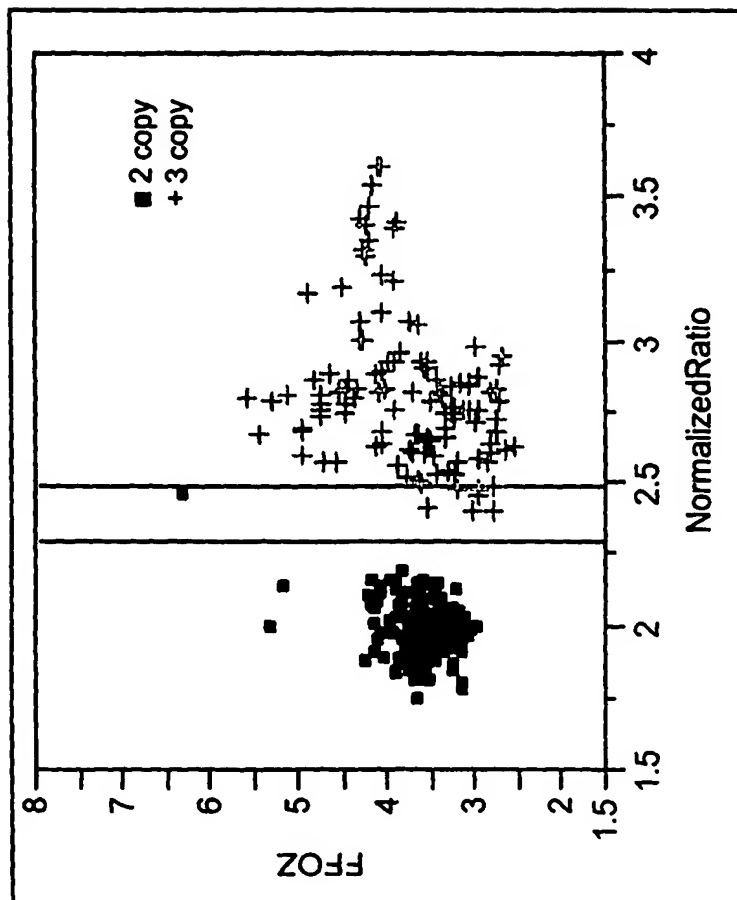
- 3 - 30 ng of DNA per reaction
- Equivocal zones: 1.4 - 1.6; 2.3 - 2.5
- N=637 samples, 517 normal, 120 aneuploid
- No call rate 1.9% (7 samples < 1.4 FOZ, 5 equivocal samples)
- Miscall rate 0%

**Figure 13B**  
**Chromosome Xq Invader Assay:MTMR8+FLJ21174 –Varying DNA Levels**



- 3 - 30 ng of DNA per reaction
- Equivocal zones: 1.4 - 1.6; 2.4 - 2.6
- N=638 samples, 518 normal, 120 aneuploid
- No call rate 3.3% (15 samples < 1.4 FOZ, 6 equivocal samples)
- Miscall rate 0%

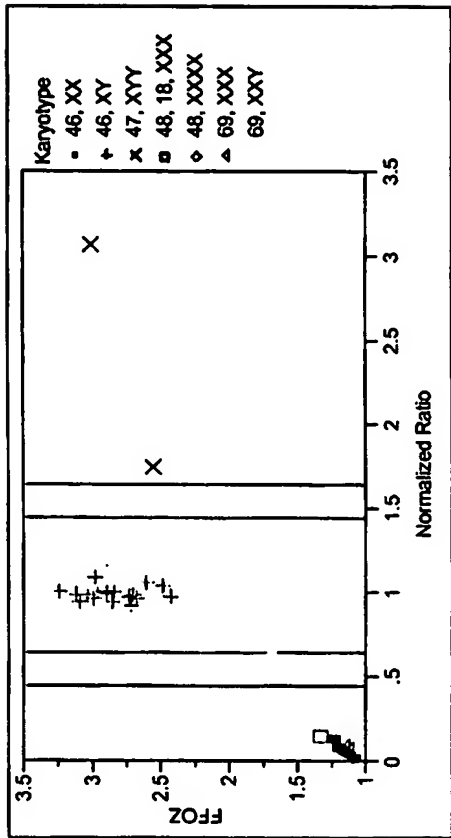
Figure 14. Chromosome 18 Invader® Assay – Sample Mixtures



- Trisomy 18 / Disomy sample mixtures
- 10 ng of DNA per reaction
- Equivocal zone: 2.3 – 2.5
- N=315 samples, 198 disomy, 117 trisomy (+18 with 0%, 10% and 20% disomy contamination)
- No call rate 2.9%
- Miscall rate 0%

Figure 15: Analysis of Triploidy Samples (69, XXY)

Normalized Ratio Analysis

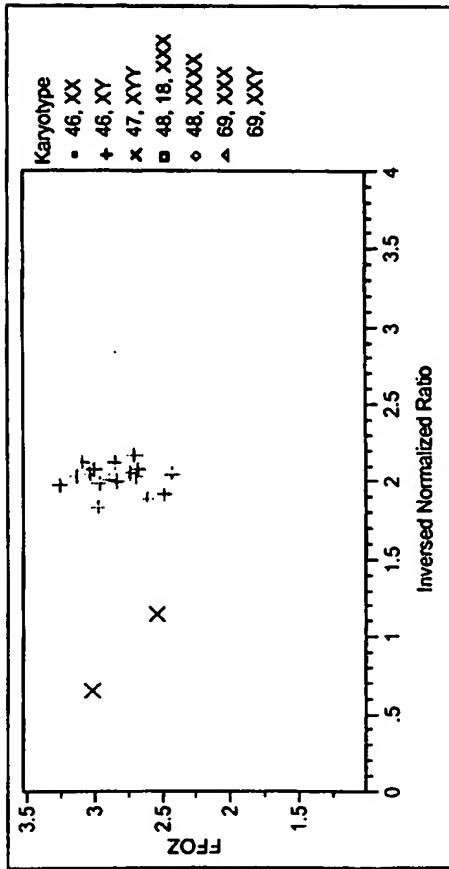


$$\text{Ratio} = \frac{(\text{Net chromosome specific FOZ})}{(\text{Net internal control FOZ})}$$

where Net FOZ = FOZ - 1

$$\text{Normalized Ratio} = \frac{(\text{Ratio of unknown sample})}{(\text{Ratio of male control sample})} \quad \text{X (1)}$$

Inversed Normalized Ratio Analysis



$$\text{Inversed Ratio} = \frac{(\text{Net internal control FOZ})}{(\text{Net chromosome specific FOZ})}$$

where Net FOZ = FOZ - 1

$$\text{Inversed Normalized Ratio} = \frac{(\text{Ratio of unknown sample})}{(\text{Ratio of male control sample})} \quad \text{X (2)}$$

Note: Samples not containing a Y chr. not shown on graph due to high inversed normalized ratios (>10)